American Journal of Medical Genetics 73:233 (1997)

Letter to the Editor

Trichorhinophalangeal Syndrome Type 2: Another Syndromic Form of Hydrometrocolpos

To the Editor:

In this journal, Mehrotra et al. [1997] reported on two sisters with the Bardet-Biedl syndrome, one of whom manifested hydrometrocolpos. Commenting on the article "Genital Abnormalities in Females with Bardet-Biedl Syndrome" by Stoler et al. [1995], they reemphasized that, "in a child with genital anomalies such as vaginal atresia and other appropriate manifestations, the diagnosis of Bardet-Biedl syndrome should come to mind."

Over the past 25 years, we have examined 18 female newborns after ultrasonographic diagnosis of hydrometrocolpos in the third trimester of pregnancy. In addition to hydrometrocolpos, three of these female newborns presented at birth with a number of associated clinical symptoms, i.e., craniofacial dysmorphism and skin and joint laxity. No specific syndromic diagnosis could be made at that age. On clinical follow up of these three female children, multiple exostoses were noted after the age of 3 years, and their distinct craniofacial appearance, with oval-shaped facies, bulbous pear-shaped nose, long prominent philtrum, thin lips, and large detached and poorly lobulated ears, was typical for the diagnosis of Langer-Giedion syndrome (Trichorhinophalangeal syndrome type 2). Prometaphase chromosome studies confirmed the presence of an interstitial deletion in the long arm of chromosome 8 in the segment 8q24.11 to 8q24.13 [Buhler et al., 1987; Fryns and Van den Berghe, 1986]. This experience indicates that high-resolution chromosome studies with special attention to the chromosomal segment 8q24.11 → 8q24.13 should be performed in all females with hydrometrocolpos if associated dysmorphic or malformative stigmata are present.

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Received 30 April 1997; Accepted 1 May 1997